



adult polyglucosan body disease

Adult polyglucosan body disease is a condition that affects the nervous system. People with this condition have problems walking due to reduced sensation in their legs (peripheral neuropathy) and progressive muscle weakness and stiffness (spasticity). Damage to the nerves that control bladder function, a condition called neurogenic bladder, causes affected individuals to have progressive difficulty controlling the flow of urine. About half of people with adult polyglucosan body disease experience a decline in intellectual function (dementia).

People with adult polyglucosan body disease typically first experience signs and symptoms related to the condition between ages 30 and 60.

Frequency

Adult polyglucosan body disease is a rare condition; although its exact prevalence is unknown, at least 70 affected individuals have been described in the medical literature.

Genetic Changes

Mutations in the *GBE1* gene cause adult polyglucosan body disease. The *GBE1* gene provides instructions for making the glycogen branching enzyme. This enzyme is involved in the production of a complex sugar called glycogen, which is a major source of stored energy in the body. Most *GBE1* gene mutations result in a shortage (deficiency) of the glycogen branching enzyme, which leads to the production of abnormal glycogen molecules. These abnormal glycogen molecules, called polyglucosan bodies, accumulate within cells and cause damage. Nerve cells (neurons) appear to be particularly vulnerable to the accumulation of polyglucosan bodies in people with this disorder, leading to impaired neuronal function.

Some mutations in the *GBE1* gene that cause adult polyglucosan body disease do not result in a shortage of glycogen branching enzyme. In people with these mutations, the activity of this enzyme is normal. How mutations cause the disease in these individuals is unclear. Other people with adult polyglucosan body disease do not have identified mutations in the *GBE1* gene. In these individuals, the cause of the disease is unknown.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- APBD
- polyglucosan body disease, adult form

Diagnosis & Management

These resources address the diagnosis or management of adult polyglucosan body disease:

- GeneReview: Adult Polyglucosan Body Disease
<https://www.ncbi.nlm.nih.gov/books/NBK5300>
- Genetic Testing Registry: Polyglucosan body disease, adult
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1849722/>
- MedlinePlus Encyclopedia: Neurogenic Bladder
<https://medlineplus.gov/ency/article/000754.htm>
- MedlinePlus Encyclopedia: Spasticity
<https://medlineplus.gov/ency/article/003297.htm>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Neurogenic Bladder
<https://medlineplus.gov/ency/article/000754.htm>
- Encyclopedia: Spasticity
<https://medlineplus.gov/ency/article/003297.htm>

- Health Topic: Dementia
<https://medlineplus.gov/dementia.html>
- Health Topic: Peripheral Nerve Disorders
<https://medlineplus.gov/peripheralnervedisorders.html>

Genetic and Rare Diseases Information Center

- Polyglucosan body disease, adult
<https://rarediseases.info.nih.gov/diseases/108/polyglucosan-body-disease-adult>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Spasticity Information Page
<https://www.ninds.nih.gov/Disorders/All-Disorders/Spasticity-Information-Page>

Educational Resources

- Boston Children's Hospital: Neurogenic Bladder
<http://www.childrenshospital.org/conditions-and-treatments/conditions/neurogenic-bladder>
- Cleveland Clinic: Dementia
<http://my.clevelandclinic.org/health/articles/types-of-dementia>
- Cleveland Clinic: Neurogenic Bladder
<http://my.clevelandclinic.org/health/articles/neurogenic-bladder>
- Cleveland Clinic: Neuropathy
<http://my.clevelandclinic.org/health/articles/neuropathy>
- Disease InfoSearch: Polyglucosan body disease, adult
<http://www.diseaseinfosearch.org/Polyglucosan+body+disease%2C+adult/5839>
- Genetic Alliance: Disease InfoSearch
<http://www.diseaseinfosearch.org/Adult+Polyglucosan+Body+Disease/250>
- JAMA Patient Page: Peripheral Neuropathy
<http://jamanetwork.com/journals/jama/fullarticle/185714>
- MalaCards: adult polyglucosan body disease
http://www.malacards.org/card/adult_polyglucosan_body_disease
- Orphanet: Adult polyglucosan body disease
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=206583
- Washington University, St. Louis: Neuromuscular Disease Center
<http://neuromuscular.wustl.edu/synmot.html#pgb>

Patient Support and Advocacy Resources

- Adult Polyglucosan Body Disease Research Foundation
<http://apbdrf.org/>
- Family Caregiver Alliance
<https://www.caregiver.org/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/adult-polyglucosan-body-disease/>

GeneReviews

- Adult Polyglucosan Body Disease
<https://www.ncbi.nlm.nih.gov/books/NBK5300>

Genetic Testing Registry

- Polyglucosan body disease, adult
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1849722/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22adult+polyglucosan+body+disease%22>

Scientific articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28adult+polyglucosan+body+disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- POLYGLUCOSAN BODY NEUROPATHY, ADULT FORM
<http://omim.org/entry/263570>

Sources for This Summary

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